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**Original Research Article**

# Clinical and etiological profile of children with pathological short stature

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## ABSTRACT

**Background:** Short stature is one of the common problem in child population with diverse aetiologies, half of them being physiological and another half due to pathological causes. The objective of this study was to study the clinical profile of the children with pathological short stature and to study the various causes for pathological stature
**Methods:** Retrospective study was conducted in the Department of Paediatrics, Chettinad Hospital and Research Institute, where the data of Children who met the inclusion criteria were collected from the case records of 2 years period from Jan 2013 to December 2014. Data collected was tabulated and analyzed using appropriate statistical methods.
**Results:** Commonest age group affected was < 5 years about 48%, 34% were 5-10 years and 17% were > 10 years. 48% were males and 52% were females. Commonest causes were Hypothyroidism and genetic disorders, followed by nutritional and musculoskeletal. 58% were proportionate and 42% were disproportionate.
**Conclusions:** Endocrinological and genetic syndromes are most common etiologies for pathological short stature. So detailed evaluation of short stature is very important as some of the pathological causes are treatable when diagnosed at an early stage.

**Keywords:** Growth, Height, Short

## INTRODUCTION

Normal growth is considered to be an indirect indicator of overall wellbeing of the child, any alteration or disturbance in the linear growth is found be one of the most common concerns for the family.

A child is considered to be short, when his or her height is below the third centile or < 2 standard deviation on the height chart used for specific population. Approximately 3% children in any population are found to be short, amongst which half of them will be physiological (familial or constitutional) and half will be pathological.[1]

Short stature is not a diagnosis but a presenting symptom, sometimes the first clinical manifestation of an underlying pathological disease, like endocrinological disease eg. Growth hormone deficiency (GHD), chronic diseases eg. Chronic renal failure, disesases of bone eg. Skeletal dysplasia or clinically defined syndromes eg. Downs; thus, short stature has to be evaluated precisely.[2]

Short stature is a result of various diverse etiologies, which are categorized in two groups broadly as pathological causes and normal variants. Normal variant short stature includes familial short stature (FSS) and constitutional growth delay (CGD), while there are broad

differential of conditions associated with pathological short stature that includes endocrine diseases, clinically defined syndromes, chronic diseases, metabolic diseases and others.[3,4]

Although after detailed evaluation, most children with short stature are assessed as being healthy careful monitoring of growth status (e.g., measurement of height, weight, and growth velocity) over time is essential for child healthcare. Most of the causes can be excluded with a thorough history, physical examination, and basic screening tests and managed accordingly as per the cause and parental counseling.

So, this study was undertaken to know the clinical and etiological profile of children who presented with pathological short stature as 50% of the cases are pathological, so that we can suspect and identify the pathological causes for short stature as early as possible and manage the condition appropriately as per the cause and counsel the parents regarding the same.

## METHODS

A retrospective study was done. Case records of children who were admitted in a tertiary care centre and diagnosed as Pathological short stature.

### *Inclusion criteria*

All Children diagnosed as short stature i.e height below 3rd centile or less than two standard deviations (SDs) below the median height for that age and sex according to the population standard, IAP growth charts were used and due to some pathological disorder being diagnosed using various investigations like, complete hemogram with erythrocyte sedimentation rate, bone age, urinalysis, stool examination, renal function test, calcium, phosphate, alkaline phosphatase, venous gas, fasting sugar, liver function tests, hormone assays like Insulin like growth factor assay, thyroid function test, karyotyping, mutation analysis and neuroimaging as per the presentation and cause in 2 years period 2013 January to 2014 December.[5]

### *Methodology*

Retrospective study was conducted in the Department of Paediatrics, Chettinad Hospital and Research Institute, where the data of children who met the inclusion criteria were collected from the case records of 2 years period from Jan 2013 to December 2014. Data obtained was arranged according to clinical profile and etiology and expressed as number and percentage and were analysed using the SPSS version 22 software. Institutional ethical committee clearance has being taken.

## RESULTS

Records of 29 children who were diagnosed as pathological short stature showed that the commonest age group being affected was < 5 years about 48% , then 34% were in the age group of 5-10 years and 17% were > 10 years (Table 1).

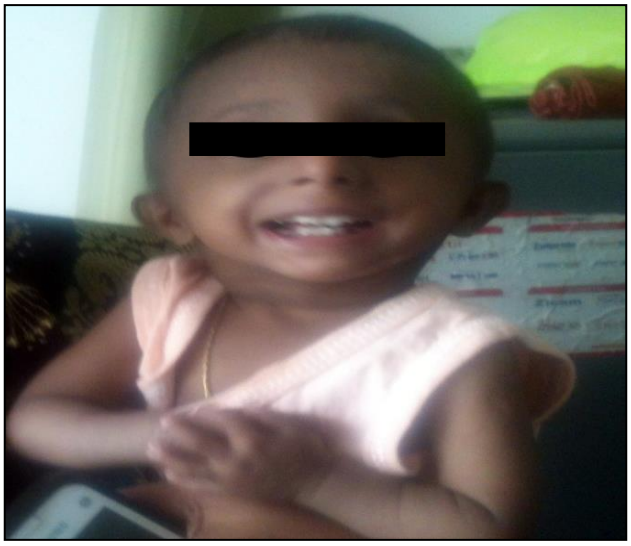

**Figure 1: Seckel syndrome.**

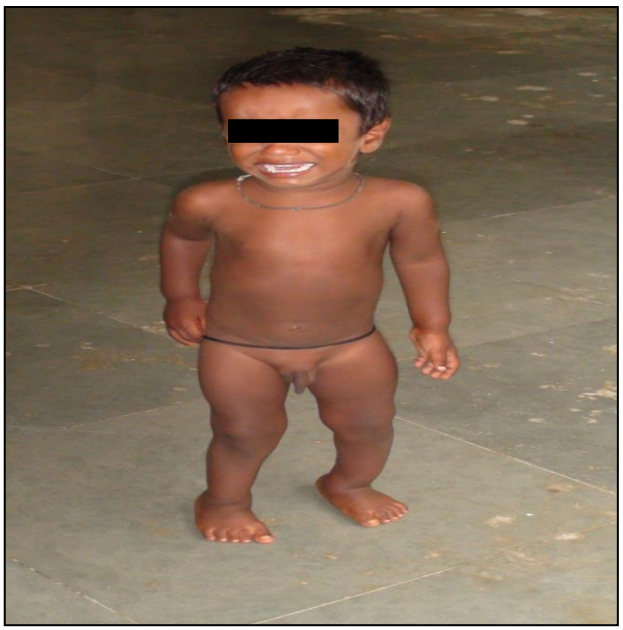

**Figure 2: Rickets.**

**Table 1: Age wise distribution.**

| Age | No (%) |
|---|---|
| 1-5 | 14 (48) |
| 6-10 | 10 (34) |
| 11-15 | 05 (17) |
| **Total** | **29 (100)** |

Among the 29 children 48% were males and 52% were females (Table 2). Analysis of various etiologies (Table 3) for the pathological short stature showed that the commonest causes were endocrinological like

hypothyroidism and genetic disorders including various syndromes like Downs, Seckel, Turners which constituted about 24% each, followed by nutritional causes and musculoskeletal causes like skeletal dysplasia, vertebral defect, MPS were about 20% each and the remaining were chronic diseases like Chronic renal failure, cyanotic congenital heart disease, caries spine and based on body proportions ,clinical profile showed 58% were in the Proportionate group and 42 % were in the Disproportionate group (Table 4).

**Table 2: Sex wise distribution.**

| Sex | No (%) |
|---|---|
| Male | 14 (48) |
| Female | 15 (52) |

**Table 3: Etiology of pathologial short stature.**

| Cause | Male (%) | Female (%) | Total (%) |
|---|---|---|---|
| Nutritional | 04 (13) | 02 (7) | 06 (20) |
| Pem | 02 (7) | 02 (7) | 04 (13) |
| Rickets | 02 (7) | 00 (0) | 02 (7) |
| Chronic diseases | 01 (3) | 02 (7) | 03 (10) |
| CCHD | | 01 (3) | 01 (3) |
| CRF | 01 (3) | | 01 (3) |
| Tb spine | | 01 (3) | 01 (3) |
| Endocrine | 04 (13) | 03 (10) | 07 (24) |
| Hypothyroidism | 03 (10) | 03 (10) | 06 (20) |
| GHD | 01 (3) | 00 (0) | 01 (3) |
| Chromosomal and genetic | 04 (13) | 03 (10) | 07 (24) |
| Downs | 01 (3) | 01 (3) | 02 (7) |
| Cornelia de lange | 00 (0) | 01 (3) | 01 (3) |
| Turners | 00 (0) | 01 (3) | 01 (3) |
| Seckel | 01 (3) | 00 (0) | 01 (3) |
| Prune belly syndrome | 01 (3) | 00 (0) | 01 (3) |
| Pierre robin sequence | 01 (3) | 00 (0) | 01 (3) |
| Musculoskeletal | 01 (3) | 05 (17) | 06 (20) |
| Skeletal dysplasia | 00 (0) | 02 (7) | 02 (7) |
| MPS | 01 (3) | 02 (7) | 03 (7) |
| Vertebral fusion defect | 00 (0) | 01 (3) | 01 (3) |
| **Total** | **14 (48)** | **15 (52)** | **29 (100)** |

**Table 4: Proportionate versus disproportionate.**

| | Proportionate: no (%) | Disproportionate: no (%) |
|---|---|---|
| **Total** | **17 (58)** | **12 (42)** |
| | • PEM: 04<br>• Chromosomal disorders: 07<br>• Rickets: 2<br>• CRF: 1<br>• CCHD: 1<br>• Vertebral fusion defect: 1 | • Hypothyroidism: 06<br>• MPS: 3<br>• Skeletal dysplasia: 2<br>• Caries spine: 1 |

And short children with presence of clinical features like with skeletal abnormalities were 31% ,with dysmorphism were 24%, with coarse, dry skin and goitre were 20%, with features of micronutrient deficiency like pallor, dermatoses, chelitis were 13% and with neonatal hypoglycaemia 3% ,with hypertension 3% ,with murmur 3% (Table 5). Among the males, nutritional, endocrinological and chronic diseases were the common etiologies and among the females skeletal causes were common though not statistically significant.

**Table 5: Clinical profile of children with pathological short stature.**

| Clinical features | Etiology | Frequency of cases | Percentage (%) |
|---|---|---|---|
| With dysmorphism | Genetic syndromes | 07 | 24 |
| With skeletal abnormalities | Skeletal dysplasia/ mucopolysaccharidosis/ caries spine/rickets | 09 | 31 |
| With dry skin/goitre | Hypothyroidism | 06 | 20 |
| With hypertension | Chronic renal failure | 01 | 3 |
| With murmur | Congenital heart disease | 01 | 3 |
| With pallor/ platynychia /dermatosis/chelitis/stomatis/phrynoderma | Protein energy malnutrition | 04 | 13 |
| With neonatal hypoglycemia/micropenis | Growth hormone defeciency | 01 | 3 |

**Table 6: Mean of chronological age and HT age for 2 most common causes.**

| | Mean HT age | Mean chronological age |
|---|---|---|
| Chromosomal disorders | 3.1 | 6.5 Years |
| Hypothyroidism | 5.8 Years | 10 Years |

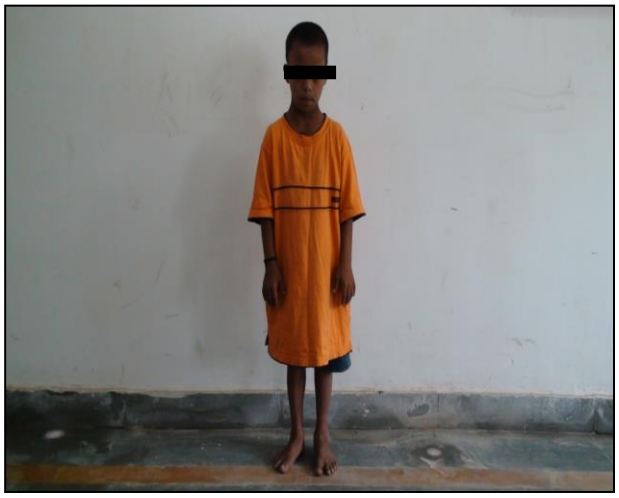

**Figure 3: Down syndrome.**

**Figure 4: Cornelia de lange syndrome.**

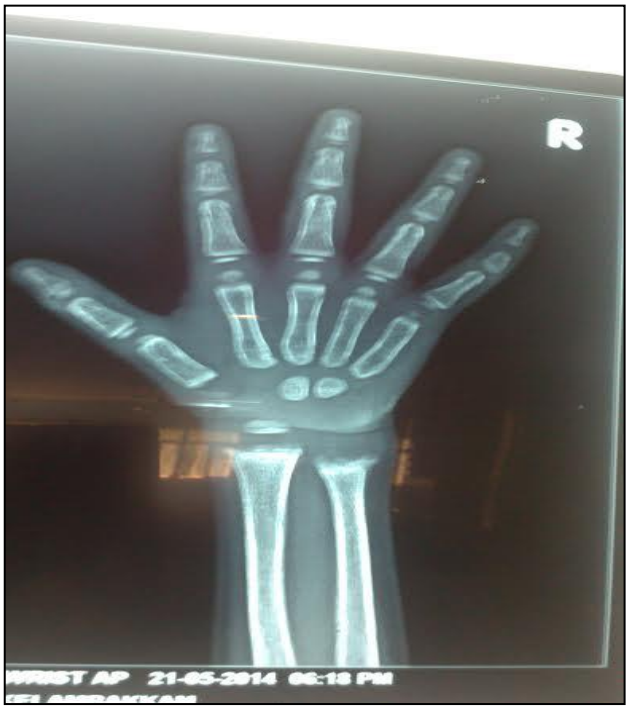


**Figure 5: X-ray wrist (6 years) in hypothyroidism.**

## DISCUSSION

In our study, the evaluation of records of 29 Children who were diagnosed as pathological short stature showed that the commonest age group being affected was < 5 years about 48%, then 34% were in the age group of 5-10 years and 17% were > 10 years. Similarly Pankaj garg in his study identified 2-5 years was most commonly affected.[6]

But this was in contrast with a study by Fahimullah etal who showed among the cases of short stature 11-15 years were the most common age affected, in another study by Rabbani MW et al the age group affected commonly was 5-11 years .[7,8] In this study, we included only those with pathological short stature, and majority of them in our study were due to endocrinological and genetic causes that presents early in life and detected early so < 5 years were more but in other two studies they included both physiological and pathological short staure. Among the 29 children 48% were males and 52% were females, this was similar to Siamak Shiva study where as various other studies showed males were commonly affected.[9] But again most of the studies included both physiological and pathological short stature.[5-12]

Analysis of various etiologies for the pathological short stature showed that the commonest causes were endocrinological like hypothyroidism and genetic disorders including various syndromes like Downs, Seckel, Turners which constituted about 24% each, followed by nutritional causes and musculoskeletal causes like skeletal dysplasia, vertebral defect, MPS were about 20% each and the remaining were chronic diseases like chronic renal failure, cyanotic congenital heart disease, caries spine and based on body proportions, 58% were in the proportionate group and 42% were in the disproportionate group. Among the males, nutritional, endocrinological and chronic diseases were the common etiologies and among the females skeletal causes were common though not statistically significant.

As said earlier most of the studies included both physiological and pathological short stature, the most common cause was physiological short stature as per various studies except a study by Pankaj Garg who showed pathological short stature was more common the etiology being Protein energy malnutrition and chronic diseases Other studies showed that among pathological short stature the commonest cause was endocrinological i.e Growth hormone deficiency and hypothyroidism. Their age ranged from 2 years and six months to 4 years. The male to female ratio was 1.3:1. The commonest etiology was genetic short stature found in 57 (51.8%) patients, while in the other 53 (48.2%) patients, variable endocrine and nutritional causes were noted.[5-12] Limitations of this study were very small sample size.

## CONCLUSION

The findings of the present study help us in developing an insight into, age-sex distribution, clinical profile (proportionate versus disproportionate), (with dysmorphic features, skeletal abnormalities, etc.) and aetiological profile of children presenting with pathological short stature.

Variety of conditions leads to short stature in children; however, in the absence of true pathology, idiopathic short stature and constitutional delay are the most frequent causes. A careful monitoring of growth and a precise evaluation of short stature are very important to identify the pathologic causes and determine whether GH treatment is needed, to reassure those who do not need treatment; and to give counselling to those who will be very short and cannot be treated.

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*Ethical approval: The study was approved by the Institutional Ethics Committee*

## REFERENCES

1. Short stature: evaluation and management. Available at www. apiindia. org/ medicine update. Accessed on 12 July 2016.
2. Khadilkar VV, Khadilkar AV. Practical approach to short stature. IJPP. 2011;13(4)396-401.
3. Ray J. Childhood short stature. Al Ameen J Med Sci. 2012;5(3):212-5.
4. Song KC, Jin SL, Kwon AR, Chae HW, Ahn JM, Kim DH. Etiologies of short stature. Ann Pediatr Endocrinol Metab. 2015;20:34-9.
5. Revised IAP growth charts, 2015. Available at iapindia.org/Revised-IAP-Growth-Charts-2015.php. Accessed on 12 July 2016.
6. Garg P. Short stature in Indian children: experience from a community level hospital. Sri Lanka J Child Health. 2004;34:84-8.
7. Ullah F. Short stature: what is the cause in our population. J Ayub Med Coll Abbottabad. 2016;28(1):135-40.
8. Rabbani MW, Khan WI, Afzal AB, Rabbani W. Causes of short stature identified in children presenting at a tertiary care hospital in Multan Pakistan. Pak J Med Sci. 2013;29(1):53-7.
9. Shiva S, Nikzad A. Etiology of short stature in East Azerbaijan, Iran. Iran J Pediatr. 2009;19(1):35-40.
10. Sultan M, Afzal M, Qureshi SM, Aziz S, Lutfullah M, Khan SA, et al. Etiology of short stature in children. J Coll Physicians Surg Pakistan. 2008;18(8):493-7.
11. Lashari SK, Korejo HB, Memon YM. To determine frequency of etiological factors in short statured patients presenting at an endocrine clinic of a tertiary care hospital. Pak J Med Sci. 2014;30(4):858-61.
12. Chowdhury SP, Sarkar TK, Haldar D, Taraphdar P, Naskar TK, Sarkar GN. Short stature in children: Experience from a tertiary care hospital in Kolkata, India. The Health. 2011;2(4):139-42.

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